

FIG. 1

## a L-periaxin

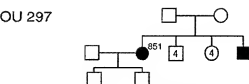
|        |   |   | POZ Domain |      |
|--------|---|---|------------|------|
| Human  | 1   | MEARS5AEELRAELVEIIVTEAQTVGSINVAGGKEGIFVRELREDSPAARSLSLQEGDQLLSARVFFENFKYEDALRLQCAEPYKVSFCLK | 97         |      |
| Murine | 1   | MEARS5AEELRAELVEIIVTEAQTVGSINVAGGKEGIFVRELREDSPAARSLSLQEGDQLLSARVFFENFKYEDALRLQCAEPYKVSFCLK | 97         |      |
| Rat    | 1   | MEARS5AEELRAELVEIIVTEAQTVGSINVAGGKEGIFVRELREDSPAARSLSLQEGDQLLSARVFFENFKYEDALRLQCAEPYKVSFCLK | 97         |      |
| Human  | RTVPTGDLLRPCTVSGYEIKGPRKAVKLVLSLSPVKKVXHVPGALGVADLPVDFESFPKESRLRGKCAAEVGVPPAAPARRKLQLPRVYRE             | NL91  | NL92       | NL93 |
| Murine | RTVPTGDLLRPCTVSGYEIKGPRKAVKLVLSLSPVKKVXHVPGALGVADLPVDFESFPKESRLRGKCAAEVGVPPAAPARRKLQLPRVYRE             |   |            |      |
| Rat    | RTVPTGDLLRPCTVSGYEIKGPRKAVKLVLSLSPVKKVXHVPGALGVADLPVDFESFPKESRLRGKCAAEVGVPPAAPARRKLQLPRVYRE             |   |            |      |
| Human  | VAEEAQAARLAAAPPRKAKVEEVAAGARFAPQVELVGPRLPGAEGVGPVSPAKPAKSAEAGFALHPTLGLGAPPAVEPAVGIQVQVEL                |   |            |      |
| Murine | VAEEAQAARLAAAPPRKAKVEEVAAGARFAPQVELVGPRLPGAEGVGPVSPAKPAKSAEAGFALHPTLGLGAPPAVEPAVGIQVQVEL                |   |            |      |
| Rat    | VAEEAQAARLAAAPPRKAKVEEVAAGARFAPQVELVGPRLPGAEGVGPVSPAKPAKSAEAGFALHPTLGLGAPPAVEPAVGIQVQVEL                |   |            |      |
| Human  | PALPSLPTLPTLCLTEGREGVSVVPTLVAAPTGVDLALPGAEEVARGEAEVALKMPRLSPFRFGARAKEVAEAKVAVSPEARVGVPLRLRPTFG          |   |            |      |
| Murine | PALPSLPTLPTLCLTEGREGVSVVPTLVAAPTGVDLALPGAEEVARGEAEVALKMPRLSPFRFGARAKEVAEAKVAVSPEARVGVPLRLRPTFG          |   |            |      |
| Rat    | PALPSLPTLPTLCLTEGREGVSVVPTLVAAPTGVDLALPGAEEVARGEAEVALKMPRLSPFRFGARAKEVAEAKVAVSPEARVGVPLRLRPTFG          |   |            |      |
| Human  | LSLLEPRPAPE-VVESKLLPTIKHPSLGTGSGVGVKPKPEVKLPKAPVKLPVPEALFEVRLPEVELPKVSEHKLKPKPENAVPEVRLPEVEL            |   |            |      |
| Murine | LSLLEPRPAPE-VVESKLLPTIKHPSLGTGSGVGVKPKPEVKLPKAPVKLPVPEALFEVRLPEVELPKVSEHKLKPKPENAVPEVRLPEVEL            |   |            |      |
| Rat    | LSLLEPRPAPE-VVESKLLPTIKHPSLGTGSGVGVKPKPEVKLPKAPVKLPVPEALFEVRLPEVELPKVSEHKLKPKPENAVPEVRLPEVEL            |   |            |      |
| Human  | PKVSEHKLKPKPENAVPEVRLPEVQLLVSEHKLKPKPENAVPEVRLPEVQLPKVSEHKLPEVSEVAVPEVRLPEVQLPKVPEHKLKPKPENAVPEVRLPEVQL |   |            |      |
| Murine | PKVSEHKLKPKPENAVPEVRLPEVQLLVSEHKLKPKPENAVPEVRLPEVQLPKVSEHKLPEVSEVAVPEVRLPEVQLPKVPEHKLKPKPENAVPEVRLPEVQL |   |            |      |
| Rat    | PKVSEHKLKPKPENAVPEVRLPEVQLLVSEHKLKPKPENAVPEVRLPEVQLPKVSEHKLPEVSEVAVPEVRLPEVQLPKVPEHKLKPKPENAVPEVRLPEVQL |   |            |      |
| Human  | EHKLEPEVQLPKV-----PENAVPDVHLEVQLPKVPEHKLKPKPENAVPDVHLEVQLPKVPEHKLKPKPENAVPEVRLPEVQLPKV                  |   |            |      |
| Murine | EHKLEPEVQLPKV-----PENAVPDVHLEVQLPKVPEHKLKPKPENAVPDVHLEVQLPKVPEHKLKPKPENAVPEVRLPEVQLPKV                  |   |            |      |
| Rat    | EHKLEPEVQLPKV-----PENAVPDVHLEVQLPKVPEHKLKPKPENAVPDVHLEVQLPKVPEHKLKPKPENAVPEVRLPEVQLPKV                  |   |            |      |
| Human  | SEHKLKPKPENAVPDVHLEVQLPKVSEHKLKPKPENAVPDVHLEVQLPKVSEHKLPEHQLPKVPEHKLKPKPENAVPEVRLPEVQLKATK              |   |            |      |
| Murine | SEHKLKPKPENAVPDVHLEVQLPKVSEHKLKPKPENAVPDVHLEVQLPKVSEHKLPEHQLPKVPEHKLKPKPENAVPEVRLPEVQLKATK              |   |            |      |
| Rat    | SEHKLKPKPENAVPDVHLEVQLPKVSEHKLKPKPENAVPDVHLEVQLPKVSEHKLPEHQLPKVPEHKLKPKPENAVPEVRLPEVQLKATK              |   |            |      |
| Human  | AEQAEQMGFEKPKPHKTHPKLGRAESPSRGGKGEAGAEVSGKLVTLFCLQPEVDGEA-HVGVPSLTPSVELDLPALGLQGGVPAKMGKGEAEQAEV        |   |            |      |
| Murine | AEQAEQMGFEKPKPHKTHPKLGRAESPSRGGKGEAGAEVSGKLVTLFCLQPEVDGEA-HVGVPSLTPSVELDLPALGLQGGVPAKMGKGEAEQAEV        |   |            |      |
| Rat    | AEQAEQMGFEKPKPHKTHPKLGRAESPSRGGKGEAGAEVSGKLVTLFCLQPEVDGEA-HVGVPSLTPSVELDLPALGLQGGVPAKMGKGEAEQAEV        |   |            |      |
| Human  | AAGVEVGRVPSVEIVTPQLPAVEIEEGLENTETKVKPSKFSFLPKFGLSGKPKVAKAEAGAGATKLKVSFKFATSLPKARVAGEAEAGGAGEAGLLP       |   |            |      |
| Murine | AAGVEVGRVPSVEIVTPQLPAVEIEEGLENTETKVKPSKFSFLPKFGLSGKPKVAKAEAGAGATKLKVSFKFATSLPKARVAGEAEAGGAGEAGLLP       |   |            |      |
| Rat    | AAGVEVGRVPSVEIVTPQLPAVEIEEGLENTETKVKPSKFSFLPKFGLSGKPKVAKAEAGAGATKLKVSFKFATSLPKARVAGEAEAGGAGEAGLLP       |   |            |      |
| Human  | ALDLSLPQLSDAHLPSGKVEVAGADLPKGPFRALPKFGVGRDTEAAELVPGVALEGGKGGGDKVPHKPKMPSFGLARGKEAVQGDRAPEGKA            |   |            |      |
| Murine | ALDLSLPQLSDAHLPSGKVEVAGADLPKGPFRALPKFGVGRDTEAAELVPGVALEGGKGGGDKVPHKPKMPSFGLARGKEAVQGDRAPEGKA            |   |            |      |
| Rat    | ALDLSLPQLSDAHLPSGKVEVAGADLPKGPFRALPKFGVGRDTEAAELVPGVALEGGKGGGDKVPHKPKMPSFGLARGKEAVQGDRAPEGKA            |   |            |      |
| Human  | ESTAVQLPKPEVLTGLGEGEAGVAVSQGLSGGKVSARQVTEGGDACLIMPPGLGSEPOVELTGFAGTGGQGAUSTVPSAEGTAGVGV                 |   |            |      |
| Murine | ESTAVQLPKPEVLTGLGEGEAGVAVSQGLSGGKVSARQVTEGGDACLIMPPGLGSEPOVELTGFAGTGGQGAUSTVPSAEGTAGVGV                 |   |            |      |
| Rat    | ESTAVQLPKPEVLTGLGEGEAGVAVSQGLSGGKVSARQVTEGGDACLIMPPGLGSEPOVELTGFAGTGGQGAUSTVPSAEGTAGVGV                 |   |            |      |
| Human  | PVNTLTPASQVAGGLLGVGVKRPITVTVQLDLDVLSKADGAATCEGGLRLKPLTLGARARVGGEGAEQPPGAERTFCLSLPDVELSPSGGN             |   |            |      |
| Murine | PVNTLTPASQVAGGLLGVGVKRPITVTVQLDLDVLSKADGAATCEGGLRLKPLTLGARARVGGEGAEQPPGAERTFCLSLPDVELSPSGGN             |   |            |      |
| Rat    | PVNTLTPASQVAGGLLGVGVKRPITVTVQLDLDVLSKADGAATCEGGLRLKPLTLGARARVGGEGAEQPPGAERTFCLSLPDVELSPSGGN             |   |            |      |
| Human  | HAETVQAEAGEGAGHKLKLVPRFLGKRAKEGAEKAKSKPLRLPVGVSQSEHVTGEGSPSPREEEEEEEGGEGAGGARGVVRVLPRVGLAAPS            |   |            |      |
| Murine | HAETVQAEAGEGAGHKLKLVPRFLGKRAKEGAEKAKSKPLRLPVGVSQSEHVTGEGSPSPREEEEEEEGGEGAGGARGVVRVLPRVGLAAPS            |   |            |      |
| Rat    | HAETVQAEAGEGAGHKLKLVPRFLGKRAKEGAEKAKSKPLRLPVGVSQSEHVTGEGSPSPREEEEEEEGGEGAGGARGVVRVLPRVGLAAPS            |   |            |      |
| Human  | KASRGDGAAPKSPVREKSKFRFRVSLSPKASGSDGEGGLRVLPVSGFSETG-----APGARMEGAGAAV                                   | 1461  |            |      |
| Murine | KASRGDGAAPKSPVREKSKFRFRVSLSPKASGSDGEGGLRVLPVSGFSETG-----APGARMEGAGAAV                                   | 1461  |            |      |
| Rat    | KASRGDGAAPKSPVREKSKFRFRVSLSPKASGSDGEGGLRVLPVSGFSETG-----APGARMEGAGAAV                                   | 1461  |            |      |

## b S-periaxin

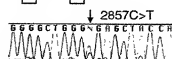
|        |   |   | POZ Domain |  |
|--------|---|---|------------|--|
| Human  | 1   | MEARS5AEELRAELVEIIVTEAQTVGSINVAGGKEGIFVRELREDSPAARSLSLQEGDQLLSARVFFENFKYEDALRLQCAEPYKVSFCLK | 97         |  |
| Murine | 1   | MEARS5AEELRAELVEIIVTEAQTVGSINVAGGKEGIFVRELREDSPAARSLSLQEGDQLLSARVFFENFKYEDALRLQCAEPYKVSFCLK | 97         |  |
| Rat    | 1   | MEARS5AEELRAELVEIIVTEAQTVGSINVAGGKEGIFVRELREDSPAARSLSLQEGDQLLSARVFFENFKYEDALRLQCAEPYKVSFCLK | 97         |  |
| Human  | RTVPTGDLLRPCTVSGYEIKGPRKAVKLVLSLSPVKKVXHVPGALGVADLPVDFESFPKESRLRGKCAAEVGVPPAAPARRKLQLPRVYRE |   |            |  |
| Murine | RTVPTGDLLRPCTVSGYEIKGPRKAVKLVLSLSPVKKVXHVPGALGVADLPVDFESFPKESRLRGKCAAEVGVPPAAPARRKLQLPRVYRE |   |            |  |
| Rat    | RTVPTGDLLRPCTVSGYEIKGPRKAVKLVLSLSPVKKVXHVPGALGVADLPVDFESFPKESRLRGKCAAEVGVPPAAPARRKLQLPRVYRE |   |            |  |

FIG. 2

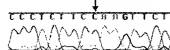
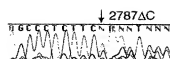
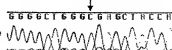
HOU 297



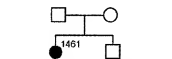
Patient 851



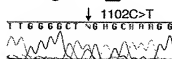
Wild type



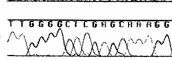
HOU 579



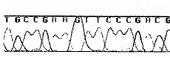
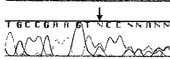
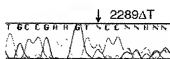
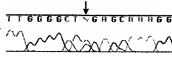
Patient 1461



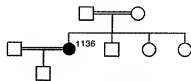
Father



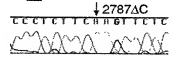
Mother



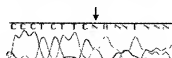
HOU 418



Patient 1136



Father



Mother

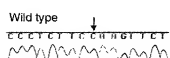
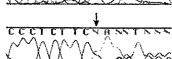
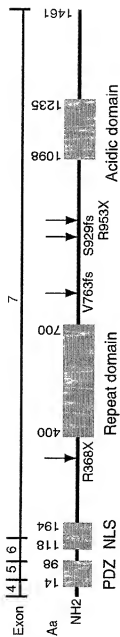


FIG. 3



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| Patient | Phenotype | Pathogenic Allele 1  | Frequency | Pathogenic Allele 2  | Frequency |
|---------|-----------|----------------------|-----------|----------------------|-----------|
| 851     | DSN       | 2857C>T<br>R953X     | 0/190     | 2787ΔC<br>S929fsX957 | 0/190     |
| 1461    | DSN       | 1102C>T<br>R368X     | 0/178     | 2289ΔT<br>V763fsX774 | 0/176     |
| 1136    | DSN       | 2787ΔC<br>S929fsX957 | 0/190     | 2787ΔC<br>S929fsX957 | 0/190     |

FIG. 4



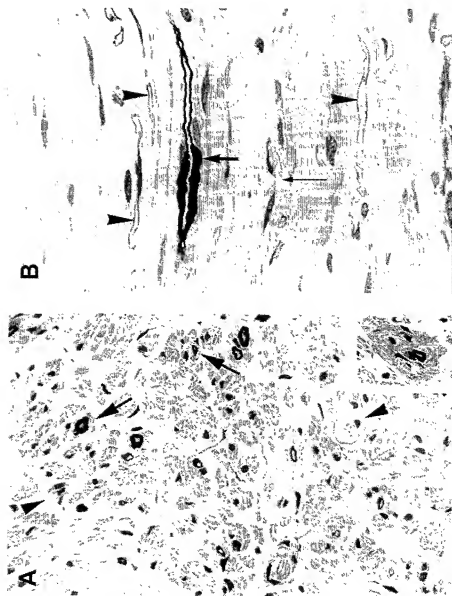


FIG. 6



FIG. 7

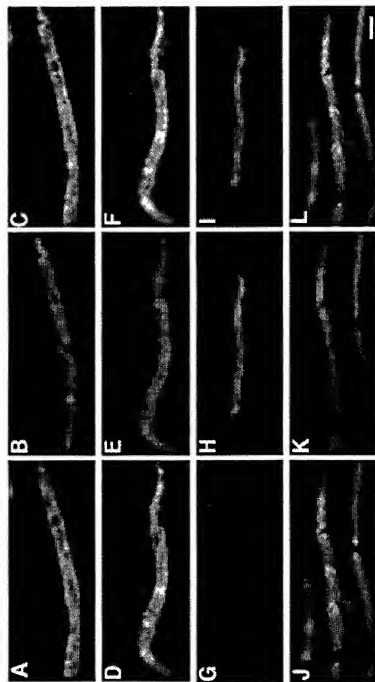


FIG. 8

FIG. 9

FIG. 9